Locus 358 (rs12202350)

SNiPAcard

Block annotations

Block info

genomic range	chr6:160,329,616-160,418,050
block size	88,435 bp
variant count	8 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.275 [-3.036 – 1.111]	gene(s) hit or close-by	IGF2R, MAS1
phastCons	μ = 0.020 [0 – 0.072]	eQTL gene(s)	IGF2R, RP11-288H12.3, SLC22A1, WTAP
GERP++	μ = 0.277 [-3.11 – 2.66]	potentially regulated gene(s)	SOD2, TCP1
CADD score	μ = 4.532 [0.785 – 13.46]	disease gene(s)	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
isobutyrylcarnitine	5.07×10^{-18}	Metabolomics GWAS Server	24816252	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SLC22A1	?	ENSG00000175003	skeletal muscle	5.46×10^{-6} (p-value)	GTEx Portal V6	3
RP11-288H12.3	?	ENSG00000213073	skeletal muscle	7.15×10^{-9} (p-value)	GTEx Portal V6	8
IGF2R	?	ENSG00000197081	skeletal muscle	3.10×10^{-38} (p-value)	GTEx Portal V6	8
IGF2R	?	ENSG00000197081	tibial nerve	3.07×10^{-9} (p-value)	GTEx Portal V6	8
RP11-288H12.3	?	ENSG00000213073	tibial nerve	1.75×10^{-5} (p-value)	GTEx Portal V6	1
IGF2R	?	ENSG00000197081	atrial appendage	4.52×10^{-9} (p-value)	GTEx Portal V6	8
IGF2R	?	ENSG00000197081	left ventricle	1.39×10^{-7} (p-value)	GTEx Portal V6	8
IGF2R	?	ENSG00000197081	subcutaneous adipocytes	1.16×10^{-9} (p-value)	GTEx Portal V6	8
WTAP	ENST00000358372	ILMN_2279339	blood	1.99×10^{-5} (p-value)	Westra et al.	1
WTAP	ENST00000621533					
IGF2R	?	ENSG00000197081	muscularis mucosae	6.22×10^{-9} (p-value)	GTEx Portal V6	3

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000468216	1	ENCP00000050519	TCP1
		ENCP00000050516	SOD2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001502170 (open chromatin region)	1	lung (IMR90)	H3K27me3
		A549	H3K27me3
		blood (K562)	H3K27me3
		skin (NHEK)	H3K27me3
		muscle (HSMM)	DNase1
ENSR00001232486 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H3K4me3, CTCF, PolII, Rad21, Jund, Sin3Ak20, ATF3, TAF1, H3K4me2, USF1, H3K9ac
		HSMMtube	H3K4me2, H3K9ac, H3K79me2, H3K27ac, DNase1
		blood (K562)	H2AZ, TAF1, PolII, H3K4me2, H3K36me3, DNase1, H3K4me3, HEY1, H3K79me2, Brg1, Egr1, H3K27ac, Max, Nfe2, E2F6, H3K9ac, USF1, Cmyc
		skin (NHDF-AD)	H3K4me1, H3K4me2, H3K9ac, DNase1, H3K4me3
		muscle (HSMM)	DNase1, H3K36me3, H3K9ac, H3K4me2, H3K27ac, H3K4me3, H3K79me2, H2AZ
		liver (HepG2)	DNase1, H3K9ac, H3K27ac, H3K4me3, H3K36me3, Gabp, PolII, H3K79me2, TAF1, ELF1, USF1, H3K4me1, H2AZ, H3K4me2
		blood (GM12878)	ATF3, PolII, H2AZ, DNase1, ELF1, H3K79me2, H3K4me3, USF1, H3K27ac, H3K4me2, H3K9ac
		lung (IMR90)	H3K9ac, H3K4me3, H3K36me3, H3K4me2, H3K79me2, H4K20me1, DNase1
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3
		skin (NHEK)	DNase1, H3K4me1, H3K27ac, H3K9ac, H3K4me2, H3K4me3, H3K36me3
		NHLF	H3K27ac, H3K9ac, H3K4me3, DNase1
		Osteobl	H2AZ, H3K27ac, H3K4me3, H3K4me2
		blood (DND-41)	H3K4me1, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	H3K4me2, H3K4me3, H3K27ac
		cervix (HeLa-S3)	DNase1, H3K36me3, PolII, H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, H3K79me2
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K27ac, H3K9ac, H4K20me1, H3K36me3, H3K27me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, DNase1, H3K27me3, PolII, H3K4me2, H3K4me3, Max
		A549	H3K4me3, H3K4me2, H3K9ac
ENSR00001232498 (promoter flanking region)	1	NHLF	DNase1
		Osteobl	H3K27ac
		blood (DND-41)	H3K36me3
		blood (K562)	PolII, Ini1, Brg1, Egr1, H3K27ac, Max, PU1, Nfe2, H3K9ac, USF1, H3K79me2, H3K4me3, H3K4me2, H3K4me1, H3K36me3, DNase1
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	H3K36me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H4K20me1, H3K36me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	DNase1, H3K79me2, H3K36me3
		blood (GM12878)	PU1, H3K79me2, H3K27ac, H3K36me3
		lung (IMR90)	H3K36me3
		nervous (NH-A)	DNase1
		A549	H3K36me3
		skin (NHEK)	DNase1, H3K4me1, H3K27ac, H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
IGF2R	upstream gene variant	1571	ENST00000356956	NM_000876.2	ENSP00000349437	1
MAS1	downstream gene variant	1213	ENST00000252660	NM_002377.2	ENSP00000252660	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
IGF2R	ENST00000356956	NM_000876.2	ENSP00000349437	4

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
MAS1	ENST00000252660	NM_002377.2	ENSP00000252660	1